Neuromuscular Disorders of Childhood Questionnaire
Australian Paediatric Surveillance Unit

Please call the APSU on (02) 9845 3005 or Dr Monique Ryan on (03) 9345 5661 if you have any questions about this form

REPORTING CLINICIANS

1. APSU Dr Code/Name: ☐☐☐☐/……. **2.** Month/Year of Report: ……../……..

PATIENT DETAILS

3. First 2 letters of first name: ☐☐ **4.** First 2 letters of surname: ☐☐

5. Date of Birth: ☐☐/☐☐/☐☐ **6.** Sex: ☐M ☐F

7. Postcode of family: ☐☐☐☐ **8.** Date of diagnosis: ☐☐/☐☐/☐☐

9. Ethnicity: ☐Aboriginal/Torres Strait Islander ☐Caucasian ☐Islander ☐Asian ☐Middle Eastern ☐African ☐Latin American ☐Indian subcontinent ☐Other Please Specify:_____

10. Mother's country of origin:_____ **11.** Father's country of origin:_____

12. Parental consanguinity? ☐ Yes ☐ No ☐ DK **If yes,** specify _____

If this patient is primarily cared for by another physician who you believe will report the case and could provide additional details, please write that physician's name in the space below then complete the questionnaire details above this line and return to APSU. If no other report is received for this child we will contact you for information requested in the remainder of the questionnaire.

The primary clinician caring for this child is: ***Name:*** ***Hospital:***

Instructions: Please answer each question by ticking the appropriate box or writing your response in the space provided. DK= Don't Know, NA = Not applicable

13. Nature of neuromuscular disorder

Please indicate the relevant diagnosis:

a. ☐ Spinal muscular atrophy; **Please specify:** ☐ Type I ☐ Type II ☐ Type III ☐ Other _____ ☐ DK

b. ☐ *Charcot-Marie-Tooth disease:
Please specify: ☐ Type I ☐ Type II ☐ Déjerine-Sottas ☐ Other _____ ☐ DK

c. ☐ Other inherited neuropathy; **Please specify:** _____

d. ☐ Chronic inflammatory demyelinating polyneuropathy

e. ☐ Congenital myasthenic syndrome; **Please specify:** _____

f. ☐ Myasthenia gravis

g. ☐ Congenital myopathy:
Please specify:
- ☐ Nemaline myopathy
- ☐ Congenital fibre-type disproportion
- ☐ Central core disease
- ☐ Multiminicore myopathy
- ☐ Myotubular myopathy
- ☐ Other, specify_____

h. ☐ Muscular dystrophy:
Please specify:
- ☐ Duchenne muscular dystrophy (DMD) ☐ Becker muscular dystrophy (BMD)
- ☐ Congenital muscular dystrophy (specify type) _____
- ☐ Facioscapulohumeral (FSH) muscular dystrophy
- ☐ Limb-girdle muscular dystrophy (specify type) _____
- ☐ Myotonic dystrophy (DM1): ☐ Congenital DM1 ☐ DM1 presenting after neonatal period

i. ☐ Dermatomyositis

j. ☐ Other inflammatory myopathy

k. ☐ Other; **Please specify:** _____

14. Presenting symptoms / signs (tick all that apply)

a. Family history ☐ Yes ☐ No ☐ DK If Yes, specify_____

b. Floppy baby ☐ Yes ☐ No ☐ DK If Yes, specify_____

c. Delayed motor milestones ☐ Yes ☐ No ☐ DK If Yes, specify_____

14. Presenting symptoms / signs (tick all that apply)

d. Abnormal gait ☐ Yes ☐ No ☐ DK If Yes, specify_____
e. Respiratory insufficiency or infection ☐ Yes ☐ No ☐ DK If Yes, specify_____
f. Orthopaedic complications ☐ Yes ☐ No ☐ DK If Yes, specify_____
g. Raised serum creatine kinase ☐ Yes ☐ No ☐ DK If Yes, specify_____
h. Cardiomyopathy or cardiac symptoms ☐ Yes ☐ No ☐ DK If Yes, specify_____
i. Cognitive deficit ☐ Yes ☐ No ☐ DK If Yes, specify_____
j. Other, specify _____

15. Current symptoms / signs (tick all that apply)

a. Delayed motor milestones ☐ Yes ☐ No ☐ DK

If yes, record age (months) achieved, or 'tick' if not yet achieved: Rolled At age _____ ☐ Not Achieved
Stood At age____ ☐ Not Achieved Walked At age____ ☐ Not Achieved

b. Abnormal gait ☐ Yes ☐ No ☐ DK If Yes, specify_____
c. Loss of independent ambulation ☐ Yes At age_____ ☐ No ☐ DK
d. Respiratory symptoms ☐ Yes ☐ No ☐ DK If Yes, specify_____
e. Orthopaedic complications ☐ Yes ☐ No ☐ DK If Yes, specify_____
f. Other, specify _____
g. Is there a family history of this disorder? ☐ Yes ☐ No ☐ DK If Yes, specify_____

16. On what basis was the diagnosis made? (tick all that apply)

a. Antenatal diagnosis ☐ Yes ☐ No ☐ DK If Yes, specify_____
b. Serum creatine kinase ☐ Normal ☐ Abnormal ☐ Not done
c. Genetic testing ☐ Yes ☐ No ☐ DK If Yes, specify_____
d Nerve conduction studies ☐ Yes ☐ No ☐ DK If Yes, specify_____
e. EMG ☐ Yes ☐ No ☐ DK If Yes, specify_____
f. Muscle or nerve biopsy ☐ Yes ☐ No ☐ DK If Yes, specify_____
g Neuroimaging ☐ Head U/S ☐ Head CT ☐ Brain MRI Specify_____ ☐ Muscle MRI
h. Other please specify _____

Service Utilisation

17. What health / educational/ community services is the child currently using?

☐ Physiotherapy ☐ Occupational therapy ☐ Speech therapy ☐ Orthotics ☐ Social work
☐ Genetic counselling ☐ Other, specify _____ ☐ DK

18. Has the family approached specific support services? ☐ Yes ☐ No ☐ DK

If Yes, which? ☐ Muscular Dystrophy Association ☐ Parent Project Australia
☐ Other, specify_____

19. Are there any services you would you like to provide for this child which are not available in your area?

_____
_____
_____

***Charcot-Marie-Tooth Data Registry**

Our research group has established a disease registry for CMT and is aiming to enroll all Australian children with CMT in this registry. Information regarding the registry will be sent to all clinicians reporting cases of CMT. Reporting a case of CMT to the APSU does not oblige you to recruit patients to the CMT Registry.

Please return this questionnaire in the addressed reply-paid envelope
Thank you for your help with this research project
Please contact the APSU on (02) 9845 3005 if you have any questions about this form